Gregory Cooper

List of Publications by Year
in descending order

86 papers	32,688 citations	61984 43 h-index	58581 82 g-index
101 all docs	101 docs citations	101 times ranked	50123 citing authors

#	Article	IF	Citations
1	Genome sequencing as a first-line diagnostic test for hospitalized infants. Genetics in Medicine, 2022, 24, 851-861.	2.4	22
2	Identifying rare, medically relevant variation via population-based genomic screening in Alabama: opportunities and pitfalls. Genetics in Medicine, 2021, 23, 280-288.	2.4	9
3	Aberrant regulation of a poison exon caused by a non-coding variant in a mouse model of Scn1a-associated epileptic encephalopathy. PLoS Genetics, 2021, 17, e1009195.	3.5	18
4	Long-read genome sequencing for the molecular diagnosis of neurodevelopmental disorders. Human Genetics and Genomics Advances, 2021, 2, 100023.	1.7	20
5	Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American Journal of Human Genetics, 2021, 108, 857-873.	6.2	19
6	Title is missing!. , 2021, 17, e1009195.		0
7	Title is missing!. , 2021, 17, e1009195.		0
8	Title is missing!. , 2021, 17, e1009195.		0
9	Title is missing!. , 2021, 17, e1009195.		0
10	Fibulin-5 mutation featuring Charcot-Marie-Tooth disease, joint hyperlaxity, and scoliosis. Neurology: Genetics, 2020, 6, e476.	1.9	0
11	A second cohort of CHD3 patients expands the molecular mechanisms known to cause Snijders Blok-Campeau syndrome. European Journal of Human Genetics, 2020, 28, 1422-1431.	2.8	25
12	De novo TBR1 variants cause a neurocognitive phenotype with ID and autistic traits: report of 25 new individuals and review of the literature. European Journal of Human Genetics, 2020, 28, 770-782.	2.8	27
13	How secondary findings are made. , 2020, , 59-75.		0
14	Non-coding and Loss-of-Function Coding Variants in TET2 are Associated with Multiple Neurodegenerative Diseases. American Journal of Human Genetics, 2020, 106, 632-645.	6.2	50
15	Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size. Brain, 2019, 142, 2617-2630.	7.6	31
16	Spatially clustering de novo variants in CYFIP2, encoding the cytoplasmic FMRP interacting protein 2, cause intellectual disability and seizures. European Journal of Human Genetics, 2019, 27, 747-759.	2.8	47
17	A YWHAZ Variant Associated With Cardiofaciocutaneous Syndrome Activates the RAF-ERK Pathway. Frontiers in Physiology, 2019, 10, 388.	2.8	23
18	Deleterious Variation in BRSK2 Associates with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 701-708.	6.2	19

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19	Child Neurology: Spastic paraparesis and dystonia with a novel ADCY5 mutation. Neurology, 2019, 93, 510-514.	1.1	7
20	Evaluating the strength of genetic results: Risks and responsibilities. PLoS Genetics, 2019, 15, e1008437.	3.5	1
21	Genome sequencing for early-onset or atypical dementia: high diagnostic yield and frequent observation of multiple contributory alleles. Journal of Physical Education and Sports Management, 2019, 5, a003491.	1.2	25
22	Clinical utility of genomic sequencing. Current Opinion in Pediatrics, 2019, 31, 732-738.	2.0	14
23	CADD: predicting the deleteriousness of variants throughout the human genome. Nucleic Acids Research, 2019, 47, D886-D894.	14.5	2,360
24	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	6.2	30
25	Systematic reanalysis of genomic data improves quality of variant interpretation. Clinical Genetics, 2018, 94, 174-178.	2.0	30
26	Genomic sequencing identifies secondary findings in a cohort of parent study participants. Genetics in Medicine, 2018, 20, 1635-1643.	2.4	24
27	Mutations in MAST1 Cause Mega-Corpus-Callosum Syndrome with Cerebellar Hypoplasia and Cortical Malformations. Neuron, 2018, 100, 1354-1368.e5.	8.1	35
28	Aberrant Inclusion of a Poison Exon Causes Dravet Syndrome and Related SCN1A-Associated Genetic Epilepsies. American Journal of Human Genetics, 2018, 103, 1022-1029.	6.2	76
29	De novo mutations in the GTP/GDP-binding region of RALA, a RAS-like small GTPase, cause intellectual disability and developmental delay. PLoS Genetics, 2018, 14, e1007671.	3.5	16
30	NBEA: Developmental disease gene with early generalized epilepsy phenotypes. Annals of Neurology, 2018, 84, 788-795.	5.3	44
31	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	3.8	46
32	Genomic diagnosis for children with intellectual disability and/or developmental delay. Genome Medicine, 2017, 9, 43.	8.2	188
33	Mutations in EBF3 Disturb Transcriptional Profiles and Cause Intellectual Disability, Ataxia, and Facial Dysmorphism. American Journal of Human Genetics, 2017, 100, 117-127.	6.2	62
34	A genome-wide interactome of DNA-associated proteins in the human liver. Genome Research, 2017, 27, 1950-1960.	5.5	10
35	A systematic comparison reveals substantial differences in chromosomal versus episomal encoding of enhancer activity. Genome Research, 2017, 27, 38-52.	5.5	244
36	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	2.5	40

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37	Clinical Sequencing Exploratory Research Consortium: Accelerating Evidence-Based Practice of Genomic Medicine. American Journal of Human Genetics, 2016, 98, 1051-1066.	6.2	137
38	De novo FGF12 mutation in 2 patients with neonatal-onset epilepsy. Neurology: Genetics, 2016, 2, e120.	1.9	29
39	Germline and somatic mutations in the MTOR gene in focal cortical dysplasia and epilepsy. Neurology: Genetics, 2016, 2, e118.	1.9	125
40	Parlez-vous VUS?. Genome Research, 2015, 25, 1423-1426.	5.5	36
41	Promoter-distal RNA polymerase II binding discriminates active from inactive CCAAT/ enhancer-binding protein beta binding sites. Genome Research, 2015, 25, 1791-1800.	5.5	30
42	Guidelines for investigating causality of sequence variants in human disease. Nature, 2014, 508, 469-476.	27.8	1,130
43	A general framework for estimating the relative pathogenicity of human genetic variants. Nature Genetics, 2014, 46, 310-315.	21.4	5,167
44	Accelerated exon evolution within primate segmental duplications. Genome Biology, 2013, 14, R9.	9.6	19
45	Distinct Properties of Cell-Type-Specific and Shared Transcription Factor Binding Sites. Molecular Cell, 2013, 52, 25-36.	9.7	283
46	Mapping genome-wide transcription factor binding sites in frozen tissues. Epigenetics and Chromatin, 2013, 6, 30.	3.9	29
47	Specifying and Sustaining Pigmentation Patterns in Domestic and Wild Cats. Science, 2012, 337, 1536-1541.	12.6	110
48	Massively parallel functional dissection of mammalian enhancers in vivo. Nature Biotechnology, 2012, 30, 265-270.	17.5	468
49	A copy number variation morbidity map of developmental delay. Nature Genetics, 2011, 43, 838-846.	21.4	1,141
50	Detection of Copy Number Variation Using SNP Genotyping. Methods in Molecular Biology, 2011, 767, 243-252.	0.9	14
51	Needles in stacks of needles: finding disease-causal variants in a wealth of genomic data. Nature Reviews Genetics, 2011, 12, 628-640.	16.3	531
52	Identification, Replication, and Functional Fine-Mapping of Expression Quantitative Trait Loci in Primary Human Liver Tissue. PLoS Genetics, 2011, 7, e1002078.	3.5	191
53	A recurrent 16p12.1 microdeletion supports a two-hit model for severe developmental delay. Nature Genetics, 2010, 42, 203-209.	21.4	539
54	Exome sequencing identifies MLL2 mutations as a cause of Kabuki syndrome. Nature Genetics, 2010, 42, 790-793.	21.4	1,238

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55	Single-nucleotide evolutionary constraint scores highlight disease-causing mutations. Nature Methods, 2010, 7, 250-251.	19.0	162
56	Targeted interrogation of copy number variation using SCIMMkit. Bioinformatics, 2010, 26, 120-122.	4.1	7
57	Evolutionary constraint facilitates interpretation of genetic variation in resequenced human genomes. Genome Research, 2010, 20, 301-310.	5.5	77
58	Identifying a High Fraction of the Human Genome to be under Selective Constraint Using GERP++. PLoS Computational Biology, 2010, 6, e1001025.	3.2	1,443
59	Recurrent reciprocal deletions and duplications of 16p13.11: the deletion is a risk factor for MR/MCA while the duplication may be a rare benign variant. Journal of Medical Genetics, 2009, 46, 223-232.	3.2	241
60	A method for rapid, targeted CNV genotyping identifies rare variants associated with neurocognitive disease. Genome Research, 2009, 19, 1579-1585.	5.5	118
61	Population Analysis of Large Copy Number Variants and Hotspots of Human Genetic Disease. American Journal of Human Genetics, 2009, 84, 148-161.	6.2	530
62	Mapping and sequencing of structural variation from eight human genomes. Nature, 2008, 453, 56-64.	27.8	983
63	Closing gaps in the human genome with fosmid resources generated from multiple individuals. Nature Genetics, 2008, 40, 96-101.	21.4	50
64	Systematic assessment of copy number variant detection via genome-wide SNP genotyping. Nature Genetics, 2008, 40, 1199-1203.	21.4	198
65	Six new loci associated with blood low-density lipoprotein cholesterol, high-density lipoprotein cholesterol or triglycerides in humans. Nature Genetics, 2008, 40, 189-197.	21.4	1,286
66	A recurrent 15q13.3 microdeletion syndrome associated with mental retardation and seizures. Nature Genetics, 2008, 40, 322-328.	21.4	509
67	Polymorphisms of the HNF1A Gene Encoding Hepatocyte Nuclear Factor-1Î± are Associated with C-Reactive Protein. American Journal of Human Genetics, 2008, 82, 1193-1201.	6.2	170
68	Rare Structural Variants Disrupt Multiple Genes in Neurodevelopmental Pathways in Schizophrenia. Science, 2008, 320, 539-543.	12.6	1,654
69	Qualifying the relationship between sequence conservation and molecular function. Genome Research, 2008, 18, 201-205.	5.5	91
70	A genome-wide scan for common genetic variants with a large influence on warfarin maintenance dose. Blood, 2008, 112, 1022-1027.	1.4	410
71	Recurrent Reciprocal Genomic Rearrangements of 17q12 Are Associated with Renal Disease, Diabetes, and Epilepsy. American Journal of Human Genetics, 2007, 81, 1057-1069.	6.2	222
72	Functional constraint and small insertions and deletions in the ENCODE regions of the human genome. Genome Biology, 2007, 8, R180.	9.6	32

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73	Analyses of deep mammalian sequence alignments and constraint predictions for 1% of the human genome. Genome Research, 2007, 17, 760-774.	5.5	184
74	Mammalian Comparative Sequence Analysis of the Agrp Locus. PLoS ONE, 2007, 2, e702.	2.5	5
75	Mutational and selective effects on copy-number variants in the human genome. Nature Genetics, 2007, 39, S22-S29.	21.4	221
76	Identification and analysis of functional elements in 1% of the human genome by the ENCODE pilot project. Nature, 2007, 447, 799-816.	27.8	4,709
77	Distribution and intensity of constraint in mammalian genomic sequence. Genome Research, 2005, 15, 901-913.	5.5	1,230
78	TRADE-OFFS IN DETECTING EVOLUTIONARILY CONSTRAINED SEQUENCE BY COMPARATIVE GENOMICS. Annual Review of Genomics and Human Genetics, 2005, 6, 143-164.	6.2	41
79	Automated Whole-Genome Multiple Alignment of Rat, Mouse, and Human. Genome Research, 2004, 14, 685-692.	5.5	79
80	Characterization of Evolutionary Rates and Constraints in Three Mammalian Genomes. Genome Research, 2004, 14, 539-548.	5.5	125
81	Genome sequence of the Brown Norway rat yields insights into mammalian evolution. Nature, 2004, 428, 493-521.	27.8	1,943
82	ABC: software for interactive browsing of genomic multiple sequence alignment data. BMC Bioinformatics, 2004, 5, 192.	2.6	12
83	Trends in occupational lead exposure since the 1978 OSHA lead standard. American Journal of Industrial Medicine, 2004, 45, 558-572.	2.1	22
84	Genomic regulatory regions: insights from comparative sequence analysis. Current Opinion in Genetics and Development, 2003, 13, 604-610.	3.3	57
85	Quantitative Estimates of Sequence Divergence for Comparative Analyses of Mammalian Genomes. Genome Research, 2003, 13, 813-820.	5.5	106
86	LAGAN and Multi-LAGAN: Efficient Tools for Large-Scale Multiple Alignment of Genomic DNA. Genome Research, 2003, 13, 721-731.	5.5	960